

Pharmacogenomics: new roles and applications for old molecules

The deepening of knowledge about the human genome is outlining a new disease taxonomy based on genetics and molecular medicine, and able to rapidly identify clinical, biochemical, molecular and functional abnormalities representative of a particular disease, thus replacing traditional phenotypic criteria that are becoming obsolete. Hence, molecular medicine changed our approach to the study of diseases in terms of diagnosis, prognosis and physiopathology, consequently influencing also the choice for the most appropriate therapy. One example is the so-called "target therapy", based on the identification of molecular targets related to functionally altered genes. Moreover, individual variability in drugs response and their different toxicity onto patients represented so far an important problem for clinical practice and for new medical treatments development. Indeed, every person can harbour genetic variants, insertions, deletions, amplifications, and about 3 millions single nucleotide polymorphisms out of 9 millions present in the genome, that are able to alter drug metabolism, response and/or toxicity, and can represent a predisposition to monofactorial genetic diseases that involve the alteration of a single gene, such as Duchenne muscular dystrophy, or to polyfactorial diseases, in which more genes are structurally and functionally relevant, as it happens for example in essential hypertension, diabetes or tumour predisposition. Molecular biology, and more generally genomic and post-genomic technologies, made it possible to recognize these alterations, and possibly will allow therapies specific for single patients, really tailored on the single cases.

Biotechnologies, by means of molecular biology techniques, gave rise to the production of human recombinant proteins that substituted those extracted from animals, such as insulin, interferon, erythropoietin, hormones and therapeutic antibodies. One of the major objectives of medical biotechnologies is to obtain for every single cellular defect a protein able to restore the right functioning, making target therapy and gene therapy concrete therapeutic options. In this period of great uncertainty of world health organizations, the increasing quantity of information on DNA sequences and the exponential development of informatics technologies, as postulated by Intell's founder Gordon Moore, will allow to convert DNA sequence information into knowledge. The discovery of DNA structure in the last century began a long journey that is near to the wire: the unification of all biological sciences under the emerging vision of biology based on information.